

# Congenital & Inherited Disorders

Division of Health Promotion & Chronic Disease Prevention

Phone: 1-800-383-3826

<http://idph.iowa.gov/genetics>



When our daughter was born, she seemed perfect—ten fingers, toes and two delightful little ears that were almost elfish. We attended the hearing screen when she was about 24 hours old and her right ear did not pass. She was screened again a month later and again did not pass. The audiologist immediately scheduled a diagnostic assessment one week later. The diagnostic assessment confirmed that she had hearing loss in her right ear. Within 22 days of being diagnosed she was fit with a hearing aid and referred to the state early intervention program. Within weeks we met with a speech pathologist and a teacher for the deaf and hard-of-hearing for evaluations and planned interventions. Ultimately, a simple hearing screen and timely follow up by Iowa's hearing screening program lead us down a path where my daughter has caught up with her peers and is at or above developmental levels for other children her age.

## Did you know?

Each year, an average of 1,850 Iowa children are born with a congenital or inherited disorder, and approximately 200 babies are stillborn. Three of every 1,000 newborns or 120 babies in Iowa are diagnosed with hearing loss each year and another 2 to 3 per 1,000 children will develop hearing loss after birth. Childhood hearing loss is the most common birth defect. Most babies born with hearing loss are born to parents with normal hearing.

The Center for Congenital and Inherited Disorders (CCID) programs serve all phases of the life cycle: prenatal, neonatal, pediatric, and adult.

## Why is the Center for Congenital and Inherited Disorders important to promoting and protecting the health of Iowans?

- Screening programs for the early detection of inherited or congenital disorders help assure earlier interventions to eliminate or reduce disability and provide family support.
- Early detection and treatment can prevent mental retardation and even death in children born with an inherited or congenital disorder.
- Children born with a hearing loss who are identified early and given appropriate intervention before 6 months of age demonstrated significantly better speech and reading comprehension than children identified after 6 months of age (Yoshinaga-Itano, et al., 1998).
- By the time a child with hearing loss graduates from high school, more than \$400,000 per child can be saved in special education costs if the child is identified early and given appropriate educational, medical, and audiological services (White & Maxon, 1995).

## Which Iowa Public Health Goals are we working to achieve?

Strengthen the public health infrastructure

Promote healthy behaviors

## What do we do?

CCID administers 8 programs that promote and improve access to comprehensive genetic health care services, laboratory services, early hearing detection and intervention, and surveillance. CCID assures statewide education is provided and develops policies and programs that assure the availability of and access to quality genetic health care, newborn screening, and laboratory services.

- Early Hearing Detection and Intervention (EHD) program – provides universal newborn hearing screening, short-term follow up, and referrals to early intervention and family support services.
- Regional Genetics Consultation Services – regional clinics provide statewide medical consultation and counseling to people with a diagnosed genetic disorder.
- Neuromuscular & Related Disorders – provides medical consultation and counseling to those with a diagnosed neuromuscular disorder, such as muscular dystrophy.
- Iowa Newborn Screening Program (INSP) – conducts newborn testing and follow-up for metabolic disorders and cystic fibrosis. Testing is done for Iowa, North Dakota, and South Dakota. INMSP also provides metabolic formula and medical foods for people diagnosed with PKU and other inherited metabolism disorders that require medically necessary foods.
- Iowa Registry for Congenital and Inherited Disorders (IRCID) – conducts surveillance for congenital and inherited disorders and stillbirth on children born in Iowa.
- Stillbirth Surveillance Program – supports stillbirth surveillance activities of the Iowa Registry for Congenital and Inherited Disorders. Promotes stillbirths awareness initiatives.
- Family Health History Initiative – provides resources to explore and compile family health history to determine the risk of inheriting disease. Provides resources for lifestyle/behavior changes and screening tests based on the results of the family health history.
- Maternal Prenatal Screening Program – conducts prenatal testing to screen for congenital/inherited disorders of the fetus.

## How do we measure our progress?

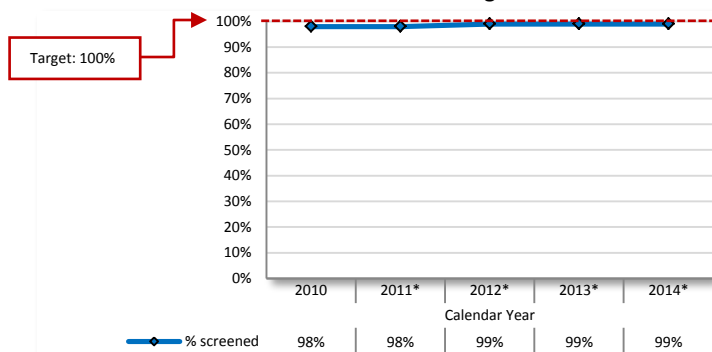
- ❶ **Percent of newborns whose screening specimens are received by the State Hygienic Laboratory within 72 hours of birth.** Data Source: INMSP/UHL database. Data are available annually.

**How are we doing?** In 2014, 99% of Iowa newborn screening specimens were received by the State Hygienic Laboratory within 72 hours of birth. (Target – 100%).

- ❷ **Percent of children, who do not have a parent-signed refusal, that are screened for disorders tested through the Iowa newborn screening panel.** Data Source: INMSP/UHL database. Data are available annually.

**How are we doing?** Nearly all, 98.72%, Iowa newborns are screened using the Iowa newborn screening panel (Target – 100%). There were 2 NBS refusals signed in CY2014.

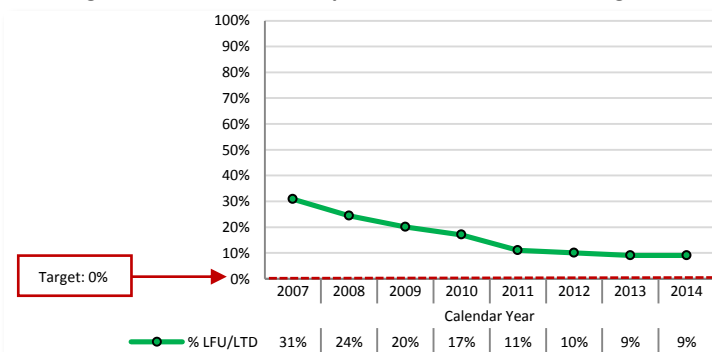
- ❸ **Percent of infants screened at birth for hearing loss.**



Data Source: IDPH/EHDI database. Data are available annually. 2014 data is preliminary.  
\*Not eligible children data removed (families who refused screening and deceased).

**How are we doing?** Nearly all Iowa newborns are screened (99%) for hearing loss. Those not eligible for screening included infant deaths and parent refusals. There were 324 families who refused the hearing screen at birth; 275 (85%) were home birth families. An additional 25 children were considered lost at birth, 21 of those were home birth families that did not respond to repeated hearing screening requests. The number of refusals slightly increased while the numbers of children lost at birth showed a significant decrease from 88 to 25 which means that education and outreach is making a difference.

- ❹ **Percent of infants lost to follow up or documentation (LFU/LTD) among all infants who did not pass their initial birth hearing screen.**



Data Source: IDPH/EHDI database. Data are available annually. 2012 data are not yet available as some children may still be receiving follow up. 2014 data is preliminary.

**How are we doing?** The number of infants that do not return for a hearing re-screen is steadily decreasing which means that a greater percentage of children are receiving recommended follow up.

## What can Iowans do to help?

- Go to <http://idph.iowa.gov/genetics> to learn about CCID programs, and <http://idph.iowa.gov/ehdi> to learn more about EHDI programs.
- Support and promote newborn screenings by having your children screened, and encouraging others to do the same.
- Conduct your own family health history and talk to your health care provider about the results.
- Talk to your legislators about funding for newborn screening and genetic programs.
- Contact the EHDI advisory committee (<http://idph.iowa.gov/ehdi/committee>) with questions or issues.

Health care professionals can

- Teach patients about the benefits of newborn screening.
- Provide information to pregnant women about monitoring fetal activity.
- Help patients gather their family health history and discuss the results with them.
- Learn more about science-based genetic research.

Policymakers can

- Learn about science-based genetic research and genetic programs.
- Provide funding for public health-based genetic programs, including public health surveillance.

## Expenditures

General fund, tobacco fund, federal funds, private grants\*, & retained fees\*: K07-0705/0709/0765; 0830-0830; 0153-0722/0724. EHDI: general fund & federal funds: K05-0611; 0153-0544/0682

	State Fiscal Year 2014 Actual	State Fiscal Year 2015 Actual	State Fiscal Year 2016 Estimate
State funds	\$956,917	\$1,037,327	\$1,162,083
Federal funds	\$521,451	\$693,985	\$651,753
Other funds*	\$284,859	\$224,869	\$506,273
<b>Total funds</b>	<b>\$1,763,226</b>	<b>\$1,956,182</b>	<b>\$2,320,109</b>
FTEs	3.24	3.78	4.10

**Note:** Funding information is intended to provide an overview of funding related to the program area. It does not include all federal and state requirements and/or restrictions for the use of funds. Contact the program area for more detailed budget information.

Iowa Department of Public Health ♦ Division of Health Promotion and Chronic Disease Prevention ♦ Congenital and Inherited Disorders

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Early Hearing Detection and Intervention program: <http://idph.iowa.gov/ehdi>